

Genetic Testing to Detect the Most Common Hereditary Cause of Retinal Hemangiomas



Why should patients with retinal hemangiomas be tested for von Hippel-Lindau Disease?

In about 30 to 60% of cases, retinal hemangiomas are associated with von Hippel-Lindau disease (VHL).^{1,2} VHL belongs to the familial cancer syndromes, which are a group of diseases characterized by a very high lifetime risk of developing certain types of tumors or cancers. It is important to identify VHL as the cause of retinal hemangioma, since patients with VHL are likely to develop additional retinal hemangiomas over time, as well as other tumors associated with VHL.¹ Once VHL has been diagnosed, annual tumor screening can help to detect tumors before they become symptomatic and allow early therapeutic intervention.³ Early diagnosis of VHL has been shown to increase survival of patients with VHL-associated renal cell carcinoma, one of the leading causes of death from VHL.⁴

Genetic testing can help to identify VHL in patients with retinal hemangioma.

VHL, like all familial cancer syndromes, is caused by a defect in a gene that is important for preventing development of certain tumors (a so-called tumor suppressor gene). Everybody carries two copies of this gene in each cell, and tumor development only occurs if both gene copies become defective in certain susceptible cells. In the general population, impairment of both gene copies in susceptible cells requires two independent events affecting the same gene (ie, one target out of a million of such targets) in the same susceptible cell (ie, one cell out of trillions of cells) – a very unlikely event. Patients with VHL, however, already carry a defect in one of their VHL-gene copies in every cell of their body. In these patients, only one additional event affecting the intact VHL-gene copy in certain susceptible cells is needed to allow tumor development from these cells – a much more likely event. Therefore, patients with VHL are at a greatly increased risk for developing the tumors associated with VHL. Genetic testing can help to diagnose VHL by detecting defects in the VHL gene.

VHL can be passed on within families.

Since children inherit their genes from their parents, they can also inherit any genetic defects. Children and siblings of a patient with VHL generally have a 50% chance of also being affected with VHL. Genetic testing can identify those family members who carry the familial VHL mutation and should undergo annual tumor screening from an early age. Conversely, genetic testing can also identify family members who do not carry the familial VHL mutation and do not need to undergo the increased tumor surveillance recommended for patients with VHL.³



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References:

1. Niemela M, Lemeta S, Sainio M, Rauma S, Pukkala E, et al (2000) Hemangioblastomas of the retina: impact of von Hippel-Lindau disease. *Invest Ophthalmol Vis Sci* 41:1909-15.
2. Singh AD, Nouri M, Shields CL, Shields JA, Smith AF (2001) Retinal capillary hemangioma: a comparison of sporadic cases and cases associated with von Hippel-Lindau disease. *Ophthalmology* 108:1907-11.
3. Sims KB (2001) Von Hippel-Lindau disease: gene to bedside. *Curr Opin Neurol* 14:695-703 Austin MA et al (2004) Familial hypercholesterolemia and coronary heart disease: a HUGE association review. *Am J Epidemiol* 160:421-429.
4. Richards FM, Webster AR, McMahon R, Woodward ER, Rose S, Maher ER (1998) Molecular genetic analysis of von Hippel-Lindau disease. *J Intern Med* 243:527-33.



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